



## SEQUENCE LISTING

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TECH CENTER 1600/2900

SubD1  
<110> Krohn, Kai  
Heino, Maarit  
Peterson, Part  
Scott S., Hamish  
Antonarakis E., Stylianos  
Lalioti D., Maria  
Shimizu, Nobuyoshi  
Kudoh, Jun

<120> NOVEL GENE DEFECTIVE IN APECED AND ITS USE

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<141> 2000-11-03

<150> PCT/FI98/00749

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100 105 110

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 Cys Val Trp Val Leu Arg Val Ser Arg Thr Trp Leu Leu Val Arg Val  
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 Heino, Maarit  
 Peterson, Part  
 Scott S., Hamish  
 Antonarakis E., Stylianos  
 Lalioti D., Maria  
 Shimizu, Nobuyoshi  
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Met Ala Thr Asp Ala Ala Leu Arg Arg Leu Leu Arg  
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Leu His Arg Thr Glu Ile Ala Val Ala Val Asp Ser Ala Phe Pro Leu  
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80 85 90

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Gln Leu Lys Ala Lys Pro Pro Lys Lys Pro Glu Ser Ser Ala Glu Gln  
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Lys Lys Cys Ile Gln Val Gly Gly Glu Phe Tyr Thr Pro Ser Lys Phe	
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Glu Asp Ser Gly Ser Gly Lys Asn Lys Ala Arg Ser Ser Ser Gly Pro	
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Gly Glu Ala Arg Leu Gly Gln Gln Gly Ser Val Pro Ala Pro Leu Ala	
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Gly Asp Val Thr Pro Ala Pro Val Glu Gly Val Leu Ala Pro Ser Pro	
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gcc cgc ctg gcc cct ggg cct gcc aag gat gac act gcc agt cac gag	1660
Ala Arg Leu Ala Pro Gly Pro Ala Lys Asp Asp Thr Ala Ser His Glu	
495 500 505	
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Pro Ala Leu His Arg Asp Asp Leu Glu Ser Leu Leu Ser Glu His Thr	
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Ala Pro Phe Pro Ser	
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Arg Asp Asp Leu Glu Ser Leu Leu Ser Glu His Thr Phe Asp Gly Ile		
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Arg	Pro	Asp	Gly	Trp	Gly	Thr	Gly	Gly	Gln	Gly	Arg	Ile	Ser	Gly	Pro		
		35				40					45						
ggc	agc	atg	gga	gca	ggg	cag	aga	ctg	ggg	agt	tca	ggg	acc	cag	aga	431	
Gly	Ser	Met	Gly	Ala	Gly	Gln	Arg	Leu	Gly	Ser	Ser	Gly	Thr	Gln	Arg		
	50				55				60					65			
tgc	tgc	tgg	ggg	agc	tgt	ttt	ggg	aag	gag	gtg	gct	ctc	agg	agg	gtg	479	
Cys	Cys	Trp	Gly	Ser	Cys	Phe	Gly	Lys	Glu	Val	Ala	Leu	Arg	Arg	Val		
				70				75					80				
ctg	cac	ccc	agc	cca	gtc	tgc	atg	ggc	gtc	tct	tgc	ctg	tgc	cag	aag	527	
Leu	His	Pro	Ser	Pro	Val	Cys	Met	Gly	Val	Ser	Cys	Leu	Cys	Gln	Lys		
			85					90					95				
aat	gag	gac	gag	tgt	gcc	gtg	tgt	cgg	gac	ggc	ggg	gag	ctc	atc	tgc	575	
Asn	Glu	Asp	Glu	Cys	Ala	Val	Cys	Arg	Asp	Gly	Gly	Glu	Leu	Ile	Cys		
		100					105					110					
tgt	gac	ggc	tgc	cct	cgg	gcc	ttc	cac	ctg	gcc	tgc	ctg	tcc	cct	ccg	623	
Cys	Asp	Gly	Cys	Pro	Arg	Ala	Phe	His	Leu	Ala	Cys	Leu	Ser	Pro	Pro		
	115					120					125						
ctc	cgg	gag	atc	ccc	agt	ggg	acc	tgg	agg	tgc	tcc	agc	tgc	ctg	cag	671	
Leu	Arg	Glu	Ile	Pro	Ser	Gly	Thr	Trp	Arg	Cys	Ser	Ser	Cys	Leu	Gln		
	130				135				140				145				
gca	aca	gtc	cag	gag	gtg	cag	ccc	cgg	gca	gag	gag	ccc	cgg	ccc	cag	719	
Ala	Thr	Val	Gln	Glu	Val	Gln	Pro	Arg	Ala	Glu	Glu	Pro	Arg	Pro	Gln		
			150					155					160				
gag	cca	ccc	gtg	gag	acc	ccg	ctc	ccc	ccg	ggg	ctt	agg	tcg	gcg	gga	767	
Glu	Pro	Pro	Val	Glu	Thr	Pro	Leu	Pro	Pro	Gly	Leu	Arg	Ser	Ala	Gly		
			165				170					175					
gag	gag	gta	aga	ggg	cca	cct	ggg	gaa	ccc	cta	gcc	ggc	atg	gac	acg	815	
Glu	Glu	Val	Arg	Gly	Pro	Pro	Gly	Glu	Pro	Leu	Ala	Gly	Met	Asp	Thr		
		180					185					190					
act	ctt	gtc	tac	aag	cac	ctg	ccg	gct	ccg	cct	tct	gca	gcc	ccg	ctg	863	

Thr	Leu	Val	Tyr	Lys	His	Leu	Pro	Ala	Pro	Pro	Ser	Ala	Ala	Pro	Leu	
195						200					205					
cca ggt ctg gac tcc tcg gcc ctg cac ccc cta ctg tgt gtg ggt cct	911															
Pro Gly Leu Asp Ser Ser Ala Leu His Pro Leu Leu Cys Val Gly Pro																
210 215 220 225																
gag ggt cag cag aac ctg gct cct ggt gcg cgt tgc ggg gtg tgc gga	959															
Glu Gly Gln Gln Asn Leu Ala Pro Gly Ala Arg Cys Gly Val Cys Gly																
230 235 240																
gat ggt acg gac gtg ctg cgg tgt act cac tgc gcc gct gcc ttc cac	1007															
Asp Gly Thr Asp Val Leu Arg Cys Thr His Cys Ala Ala Ala Phe His																
245 250 255																
tgg cgc tgc cac ttc cca gcc ggc acc tcc cgg ccc ggg acg ggc ctg	1055															
Trp Arg Cys His Phe Pro Ala Gly Thr Ser Arg Pro Gly Thr Gly Leu																
260 265 270																
cgc tgc aga tcc tgc tca gga gac gtg acc cca gcc cct gtg gag ggg	1103															
Arg Cys Arg Ser Cys Ser Gly Asp Val Thr Pro Ala Pro Val Glu Gly																
275 280 285																
gtg ctg gcc ccc agc ccc gcc cgc ctg gcc cct ggg cct gcc aag gat	1151															
Val Leu Ala Pro Ser Pro Ala Arg Leu Ala Pro Gly Pro Ala Lys Asp																
290 295 300 305																
gac act gcc agt cac gag ccc gct ctg cac agg gat gac ctg gag tcc	1199															
Asp Thr Ala Ser His Glu Pro Ala Leu His Arg Asp Asp Leu Glu Ser																
310 315 320																
ctt ctg agc gag cac acc ttc gat ggc atc ctg cag tgg gcc atc cag	1247															
Leu Leu Ser Glu His Thr Phe Asp Gly Ile Leu Gln Trp Ala Ile Gln																
325 330 335																
agc atg gcc cgt ccg gcg gcc ccc ttc ccc tcc tga cccagatgg	1293															
Ser Met Ala Arg Pro Ala Ala Pro Phe Pro Ser																
340 345																
ccgggacatg cagctctgat gagagagtgc tgagaaggac acctccttcc tcagtcctgg	1353															
aagccggccg gctgggatca agaaggggac agcgccacct cttgtcagtg ctcggctgta	1413															
aacagctctg tgtttctggg gacaccagcc atcatgtgcc tggaaattaa accctgcccc	1473															
acttctctac tctggaagtc cccgggagcc tctccttgcc tggtgaccta ctaaaaatat	1533															
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<211> 348

<212> PRT

<213> Homo sapiens

<400> 4

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Arg Asn Arg Val Phe Phe Pro Ile Gly Met Ala Pro Gly Gly Val Cys  
20 25 30

Ser Arg Pro Asp Gly Trp Gly Thr Gly Gly Gln Gly Arg Ile Ser Gly  
35 40 45

Pro Gly Ser Met Gly Ala Gly Gln Arg Leu Gly Ser Ser Gly Thr Gln  
50 55 60

Arg Cys Cys Trp Gly Ser Cys Phe Gly Lys Glu Val Ala Leu Arg Arg  
65 70 75 80

Val Leu His Pro Ser Pro Val Cys Met Gly Val Ser Cys Leu Cys Gln  
85 90 95

Lys Asn Glu Asp Glu Cys Ala Val Cys Arg Asp Gly Gly Glu Leu Ile  
100 105 110

Cys Cys Asp Gly Cys Pro Arg Ala Phe His Leu Ala Cys Leu Ser Pro  
115 120 125

Pro Leu Arg Glu Ile Pro Ser Gly Thr Trp Arg Cys Ser Ser Cys Leu  
130 135 140

Gln Ala Thr Val Gln Glu Val Gln Pro Arg Ala Glu Glu Pro Arg Pro  
145 150 155 160

Gln Glu Pro Pro Val Glu Thr Pro Leu Pro Pro Gly Leu Arg Ser Ala  
165 170 175

Gly Glu Glu Val Arg Gly Pro Pro Gly Glu Pro Leu Ala Gly Met Asp  
180 185 190

Thr Thr Leu Val Tyr Lys His Leu Pro Ala Pro Pro Ser Ala Ala Pro  
195 200 205

Leu Pro Gly Leu Asp Ser Ser Ala Leu His Pro Leu Leu Cys Val Gly



210	215	220
Pro Glu Gly Gln Gln Asn Leu Ala Pro Gly Ala Arg Cys Gly Val Cys		
225	230	235 240
Gly Asp Gly Thr Asp Val Leu Arg Cys Thr His Cys Ala Ala Ala Phe		
	245	250 255
His Trp Arg Cys His Phe Pro Ala Gly Thr Ser Arg Pro Gly Thr Gly		
	260	265 270
Leu Arg Cys Arg Ser Cys Ser Gly Asp Val Thr Pro Ala Pro Val Glu		
	275	280 285
Gly Val Leu Ala Pro Ser Pro Ala Arg Leu Ala Pro Gly Pro Ala Lys		
	290	295 300
Asp Asp Thr Ala Ser His Glu Pro Ala Leu His Arg Asp Asp Leu Glu		
305	310	315 320
Ser Leu Leu Ser Glu His Thr Phe Asp Gly Ile Leu Gln Trp Ala Ile		
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Gln Ser Met Ala Arg Pro Ala Ala Pro Phe Pro Ser		
	340	345

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 <212> DNA  
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 atccactggg aatgccatgc tcattcttcg tccccagcat gggtttcttaa tggggtagaa 180

gcaggtcggg agagacctcc ctgggcctgg ccccactgcc ctgtgaggaa gggttc atg 239  
Met  
1

tgg ttg gtg tac agt tcc ggg gcc cct gga acg cag cag cct gca aga 287  
Trp Leu Val Tyr Ser Ser Gly Ala Pro Gly Thr Gln Gln Pro Ala Arg  
5 10 15

aac cgg gtt ttc ttc cca ata ggg atg gcc ccg ggg ggt gtc tgt tgg 335  
Asn Arg Val Phe Phe Pro Ile Gly Met Ala Pro Gly Gly Val Cys Trp  
20 25 30

aga cca gat gga tgg gga aca ggt ggt cag ggc aga att tca ggc cct 383  
Arg Pro Asp Gly Trp Gly Thr Gly Gly Gln Gly Arg Ile Ser Gly Pro  
35 40 45

ggc agc atg gga gca ggg cag aga ctg ggg agt tca ggt acc cag aga 431  
Gly Ser Met Gly Ala Gly Gln Arg Leu Gly Ser Ser Gly Thr Gln Arg  
50 55 60 65

tgc tgc tgg ggg agc tgt ttt ggg aag gag gtg gct ctc agg agg gtg 479  
Cys Cys Trp Gly Ser Cys Phe Gly Lys Glu Val Ala Leu Arg Arg Val  
70 75 80

ctg cac ccc agc cca gtc tgc atg ggc gtc tct tgc ctg tgc cag aag 527  
Leu His Pro Ser Pro Val Cys Met Gly Val Ser Cys Leu Cys Gln Lys  
85 90 95

aat gag gac gag tgt gcc gtg tgt cgg gac ggc ggg gag ctc atc tgc 575  
Asn Glu Asp Glu Cys Ala Val Cys Arg Asp Gly Gly Glu Leu Ile Cys  
100 105 110

tgt gac ggc tgc cct cgg gcc ttc cac ctg gcc tgc ctg tcc cct ccg 623  
Cys Asp Gly Cys Pro Arg Ala Phe His Leu Ala Cys Leu Ser Pro Pro  
115 120 125

ctc cgg gag atc ccc agt ggg acc tgg agg tgc tcc agc tgc ctg cag 671  
Leu Arg Glu Ile Pro Ser Gly Thr Trp Arg Cys Ser Ser Cys Leu Gln  
130 135 140 145

gca aca gtc cag gag gtg cag ccc cgg gca gag gag ccc cgg ccc cag 719  
Ala Thr Val Gln Glu Val Gln Pro Arg Ala Glu Glu Pro Arg Pro Gln  
150 155 160

gag cca ccc gtg gag acc ccg ctc ccc ccg ggg ctt agg tgc gcg gga 767  
Glu Pro Pro Val Glu Thr Pro Leu Pro Pro Gly Leu Arg Ser Ala Gly  
165 170 175

gag gag ccc cgc tgc cag ggc tgg act cct cgg ccc tgc acc ccc tac 815  
 Glu Glu Pro Arg Cys Gln Gly Trp Thr Pro Arg Pro Cys Thr Pro Tyr  
 180 185 190

tgt gtg tgg gtc ctg agg gtc agc aga acc tgg ctc ctg gtg cgc gtt 863  
 Cys Val Trp Val Leu Arg Val Ser Arg Thr Trp Leu Leu Val Arg Val  
 195 200 205

gcg ggg tgt gcg gag atg gta cgg acg tgc tgc ggt gta ctc act gcg 911  
 Ala Gly Cys Ala Glu Met Val Arg Thr Cys Cys Gly Val Leu Thr Ala  
 210 215 220 225

ccg ctg cct tcc act ggc gct gcc act tcc cag ccg gca cct ccc ggc 959  
 Pro Leu Pro Ser Thr Gly Ala Ala Thr Ser Gln Pro Ala Pro Pro Gly  
 230 235 240

ccg gga cgg gcc tgc gct gca gat cct gct cag gag acg tga 1001  
 Pro Gly Arg Ala Cys Ala Ala Asp Pro Ala Gln Glu Thr  
 245 250 255

cccagcccc tgtggagggg gtgctggccc ccagccccgc ccgcctggcc cctgggcctg 1061  
 ccaaggatga cactgccagt cacgagcccg ctctgcacag ggatgacctg gagtcccttc 1121  
 tgagcgagca caccttcgat ggcatactgc agtggggccat ccagagcatg gcccgccgg 1181  
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 agcgccacct cttgtcagtg ctcggtgta aacagctctg tgtttctggg gacaccagcc 1361  
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<212> PRT

<213> Homo sapiens

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Arg Asn Arg Val Phe Phe Pro Ile Gly Met Ala Pro Gly Gly Val Cys

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Trp	Arg	Pro	Asp	Gly	Trp	Gly	Thr	Gly	Gly	Gln	Gly	Arg	Ile	Ser	Gly		
	35					40					45						
Pro	Gly	Ser	Met	Gly	Ala	Gly	Gln	Arg	Leu	Gly	Ser	Ser	Gly	Thr	Gln		
	50					55					60						
Arg	Cys	Cys	Trp	Gly	Ser	Cys	Phe	Gly	Lys	Glu	Val	Ala	Leu	Arg	Arg		
65					70					75					80		
Val	Leu	His	Pro	Ser	Pro	Val	Cys	Met	Gly	Val	Ser	Cys	Leu	Cys	Gln		
				85					90						95		
Lys	Asn	Glu	Asp	Glu	Cys	Ala	Val	Cys	Arg	Asp	Gly	Gly	Glu	Leu	Ile		
			100						105				110				
Cys	Cys	Asp	Gly	Cys	Pro	Arg	Ala	Phe	His	Leu	Ala	Cys	Leu	Ser	Pro		
		115					120					125					
Pro	Leu	Arg	Glu	Ile	Pro	Ser	Gly	Thr	Trp	Arg	Cys	Ser	Ser	Cys	Leu		
	130					135					140						
Gln	Ala	Thr	Val	Gln	Glu	Val	Gln	Pro	Arg	Ala	Glu	Glu	Pro	Arg	Pro		
145					150					155					160		
Gln	Glu	Pro	Pro	Val	Glu	Thr	Pro	Leu	Pro	Pro	Gly	Leu	Arg	Ser	Ala		
				165					170					175			
Gly	Glu	Glu	Pro	Arg	Cys	Gln	Gly	Trp	Thr	Pro	Arg	Pro	Cys	Thr	Pro		
			180					185					190				
Tyr	Cys	Val	Trp	Val	Leu	Arg	Val	Ser	Arg	Thr	Trp	Leu	Leu	Val	Arg		
	195						200					205					
Val	Ala	Gly	Cys	Ala	Glu	Met	Val	Arg	Thr	Cys	Cys	Gly	Val	Leu	Thr		
	210					215					220						
Ala	Pro	Leu	Pro	Ser	Thr	Gly	Ala	Ala	Thr	Ser	Gln	Pro	Ala	Pro	Pro		
225					230					235				240			
Gly	Pro	Gly	Arg	Ala	Cys	Ala	Ala	Asp	Pro	Ala	Gln	Glu	Thr				
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<213> Artificial Sequence

<220>

<223> Description of Artificial Sequence: PRIMER

<400> 7

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20

<210> 8

<211> 22

<212> DNA

<213> Artificial Sequence

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<223> Description of Artificial Sequence: PRIMER

<400> 8

gttcccagagt ggaaggcgct gc

22

<210> 9

<211> 20

<212> DNA

<213> Artificial Sequence

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<223> Description of Artificial Sequence: PRIMER

<400> 9

aggggacagg caggccaggt

20

<210> 10

<211> 24

<212> DNA

<213> Artificial Sequence

<220>

<223> Description of Artificial Sequence: PRIMER

<400> 10

gagttcaggt acccagagat gctg

24

<210> 11

<211> 20

<212> DNA

<213> Artificial Sequence

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<223> Description of Artificial Sequence: PRIMER

<400> 11

ctcgctcaga agggactcca

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<210> 12

<211> 23

<212> DNA

<213> Artificial Sequence

<220>

<223> Description of Artificial Sequence: PRIMER

<400> 12

ggattcagac catgtcagct tca

23

<210> 13

<211> 21

<212> DNA

<213> Artificial Sequence

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<223> Description of Artificial Sequence: PRIMER

<400> 13

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21

<210> 14

<211> 21

<212> DNA

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<223> Description of Artificial Sequence: PRIMER

<400> 14

tggatgagga tcccctccac g

21

<210> 15

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<400> 19  
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<210> 20  
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<400> 20  
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<210> 21  
<211> 24  
<212> DNA  
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<400> 21  
tggagatggg caggccgcag ggtg 24

<210> 22  
<211> 24  
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<223> Description of Artificial Sequence: PRIMER

<400> 22  
cagtccagct gggctgagca ggtg 24

<210> 23  
<211> 24  
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<213> Artificial Sequence

<220>

<223> Description of Artificial Sequence: PRIMER

<400> 23

gcggctccaa gaagtgcac cagg

24

<210> 24

<211> 24

<212> DNA

<213> Artificial Sequence

<220>

<223> Description of Artificial Sequence: PRIMER

<400> 24

ctccaccctg caaggaagag gggc

24

<210> 25

<211> 16

<212> PRT

<213> Artificial Sequence

<220>

<223> Description of Artificial Sequence: PRIMER

<400> 25

Thr	Leu	His	Leu	Lys	Glu	Lys	Glu	Gly	Cys	Pro	Val	Gln	Ala	Phe	His
1				5				10						15	

<210> 26

<211> 15

<212> PRT

<213> Artificial Sequence

<220>

<223> Description of Artificial Sequence: PRIMER

<400> 26

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<220>  
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<400> 27  
atggcgacgg acgcggcgct acgc 24

<210> 28  
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<223> Description of Artificial Sequence: PRIMER

<400> 28  
cctggatgta cttcttgag ccgc 24

<210> 29  
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<223> Description of Artificial Sequence: PRIMER

<400> 29  
gagcccgagg ggccgtggag ggga 24

<210> 30  
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<212> DNA  
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<220>  
<223> Description of Artificial Sequence: PRIMER

<400> 30  
ggctgcacct cctggactgt tgcc 24

<210> 31

<211> 24  
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 <210> 32  
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<211> 24  
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<223> Description of Artificial Sequence: PRIMER

<400> 35

ggactgagga aggaggtgtc cttc

24

<210> 36

<211> 20

<212> PRT

<213> Artificial Sequence

<220>

<223> Description of Artificial Sequence: PRIMER

<400> 36

Asp Gly Ile Leu Gln Trp Ala Ile Gln Ser Met Ala Arg Pro Ala Ala  
1 5 10 15

Pro Phe Pro Ser  
20